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Survey Supports Idea That Familial Chiari Linked to CTDs

By now, it's well recognized that sometimes Chiari runs in families. According to the Chiari1000, 12% of patients have an immediate family member who has also been diagnosed. However, the mechanism by which this occurs is not known and Chiari genetic studies have provided limited clues. One idea is that it has to do with connective tissue disorders, such as hypermobile Ehlers-Danlos Syndrome (hEDS), which are known to be heritable. Since a sub-group of Chiari patients are known to also have hEDS, and connective tissue disorders are heritable, then logically that could explain at least a portion of the familial Chiari cases.

To investigate this, researchers sent a survey to all surgical patients who had been seen at their facility over a 15-year period. The short survey was comprised of 10 questions which asked if other family members had been diagnosed with Chiari and whether there was a family history of symptoms and conditions related to connective tissue disorders, such as joint injuries and easy bruising. Out of 890 patients, the research team received 354 complete responses. Among the respondents, 87% reported a family history of Chiari, a rate far higher than has been reported in the general Chiari population, suggesting the survey disproportionately attracted families already aware of multiple affected relatives.

Despite this, the researchers split the respondents into two groups, familial and sporadic (no family history of Chiari) in order to statistically compare their responses to the other questions. They found that the familial group had significantly higher rates of a family history of joint issues such as dislocations, injuries, and replacements. The familial group also reported higher rates of easy bruising in their family, along with higher family rates of diagnosed connective tissue disorders. Finally, the familial group reported more hypermobility than the sporadic group.

One aspect of the responses that the authors did not address is that even among the sporadic group, the rates of family history of easy bruising (57%) and hypermobility (48%) were very high. The implications of this are not clear.

It is important to note the methodological limitations of this study. The responses were heavily skewed in favor of familial Chiari cases. In addition, the survey questions were not very specific in terms of defining "easy bruising" and "hyperflexibility". Both factors limit the interpretation and generalization of the results. However, this study is a good first step in investigating a topic that is of great interest to the Chiari community.

Source: Heukwa-Tefoung A, Bui A, Gilmer H. Familial Chiari Malformation: Prevalence of Connective Tissue Disorders and Other Comorbidities. Neurosurgery. 2026 Jul 22. doi: 10.1227/neu.0000000000004172. Epub ahead of print. PMID: 42484321.

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